



<110> HEINO, Kai

HEINO, Maarit

PETERSON, Part

SCOTT, Hamish

ANTONARAKIS, Stylianos

LALIOTI, Maria D.

SHIMIZU, Nobuyoshi D.

KUDOH, Jun D.

<120> NOVEL GENE DEFECTIVE IN APECED AND ITS USE

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<140> 09/508,658

<141> 2000-11-03

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<170> PatentIn version 3.2

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Leu His Arg Thr Glu Ile Ala Val Ala Val Asp Ser Ala Phe Pro Leu

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Glu	Thr	Leu	His	Leu	Lys	Glu	Lys	Glu	Gly	Cys	Pro	Gln	Ala	Phe	His		
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Ala	Leu	Leu	Ser	Trp	Leu	Leu	Thr	Gln	Asp	Ser	Thr	Ala	Ile	Leu	Asp		
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Pro	Pro	Arg	Leu	Pro	Thr	Lys	Arg	Lys	Ala	Ser	Glu	Glu	Ala	Arg	Ala		
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Ala Pro Gly Ala Arg Cys Gly Val Cys Gly Asp Gly Thr Asp Val Leu	
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Arg Cys Thr His Cys Ala Ala Ala Phe His Trp Arg Cys His Phe Pro	
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Ala Gly Thr Ser Arg Pro Gly Thr Gly Leu Arg Cys Arg Ser Cys Ser	

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Pro Ala Leu His Arg Asp Asp Leu Glu Ser Leu Leu Ser Glu His Thr			
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Pro Gln Leu His Gln Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp  
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Arg Cys Gly Val Cys Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His  
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Cys Ala Ala Ala Phe His Trp Arg Cys His Phe Pro Ala Gly Thr Ser  
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 Trp Leu Val Tyr Ser Ser Gly Ala Pro Gly Thr Gln Gln Pro Ala Arg  
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Asp Thr Ala Ser His Glu Pro Ala Leu His Arg Asp Asp Leu Glu Ser			
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Pro Gly Ser Met Gly Ala Gly Gln Arg Leu Gly Ser Ser Gly Thr Gln
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Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile  
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Gly Glu Glu Val Arg Gly Pro Pro Gly Glu Pro Leu Ala Gly Met Asp  
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Thr Thr Leu Val Tyr Lys His Leu Pro Ala Pro Pro Ser Ala Ala Pro  
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Pro Glu Gly Gln Gln Asn Leu Ala Pro Gly Ala Arg Cys Gly Val Cys  
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Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His Cys Ala Ala Ala Phe  
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35 40 45

Pro Gly Ser Met Gly Ala Gly Gln Arg Leu Gly Ser Ser Gly Thr Gln
50 55 60

Arg Cys Cys Trp Gly Ser Cys Phe Gly Lys Glu Val Ala Leu Arg Arg
65 70 75 80

Val Leu His Pro Ser Pro Val Cys Met Gly Val Ser Cys Leu Cys Gln
85 90 95

Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile
100 105 110

Cys Cys Asp Gly Cys Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro  
115 120 125

Pro Leu Arg Glu Ile Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys Leu  
130 135 140

Gln Ala Thr Val Gln Glu Val Gln Pro Arg Ala Glu Glu Pro Arg Pro  
145 150 155 160

Gln Glu Pro Pro Val Glu Thr Pro Leu Pro Pro Gly Leu Arg Ser Ala  
165 170 175

Gly Glu Glu Pro Arg Cys Gln Gly Trp Thr Pro Arg Pro Cys Thr Pro  
180 185 190

Tyr Cys Val Trp Val Leu Arg Val Ser Arg Thr Trp Leu Leu Val Arg  
195 200 205

Val Ala Gly Cys Ala Glu Met Val Arg Thr Cys Cys Gly Val Leu Thr  
210 215 220

Ala Pro Leu Pro Ser Thr Gly Ala Ala Thr Ser Gln Pro Ala Pro Pro  
225 230 235 240

Gly Pro Gly Arg Ala Cys Ala Ala Asp Pro Ala Gln Glu Thr  
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20

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<210> 18  
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24

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Thr Leu His Leu Lys Glu Lys Glu Gly Cys Pro Val Gln Ala Phe His  
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<210> 26  
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<400> 33  
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<400> 34  
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<210> 35  
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<400> 35

ggactgagga aggaggtgtc cttc

24

<210> 36

<211> 20

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<400> 36

Asp Gly Ile Leu Gln Trp Ala Ile Gln Ser Met Ala Arg Pro Ala Ala  
1 5 10 15

Pro Phe Pro Ser  
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<210> 37

<211> 42

<212> PRT

<213> Homo sapiens

<400> 37

Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile Cys Cys Asp Gly Cys  
1 5 10 15

Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro Pro Leu Arg Glu Ile  
20 25 30

Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys  
35 40

<210> 38

<211> 42

<212> PRT

<213> HOMO SAPIENS

<400> 38

Cys Gly Val Cys Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His Cys  
1 5 10 15

Ala Ala Ala Phe His Trp Arg Cys His Phe Pro Ala Gly Thr Ser Arg  
20 25 30

Pro Gly Thr Gly Leu Arg Cys Arg Ser Cys  
35 40

<210> 39  
<211> 42  
<212> PRT  
<213> HOMO SAPIENS

<400> 39

Cys Glu Val Cys Gln Gln Gly Gly Glu Ile Ile Leu Cys Asp Thr Cys  
1 5 10 15

Pro Arg Ala Thr His Met Val Cys Leu Asp Pro Asp Met Glu Lys Ala  
20 25 30

Pro Glu Gly Leu Trp Ser Cys Pro His Cys  
35 40

<210> 40  
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<212> PRT  
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<400> 40

Cys Arg Val Cys Lys Asp Gly Gly Glu Leu Ile Cys Cys Asp Thr Cys  
1 5 10 15

Pro Ser Ser Tyr His Ile His Cys Leu Asn Pro Pro Leu Pro Glu Ile  
20 25 30

Pro Asn Gly Glu Trp Leu Cys Pro Arg Cys  
35 40

<210> 41  
<211> 42  
<212> PRT  
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<400> 41

Cys Ala Val Cys Gln Asn Gly Gly Glu Leu Ile Cys Cys Glu Lys Cys  
1 5 10 15

Pro Lys Val Phe His Leu Ser Cys His Val Pro Thr Leu Thr Asn Phe  
20 25 30

Pro Ser Gly Glu Trp Ile Cys Thr Phe Cys  
35 40